

# Familial hypercholesterolaemia overview

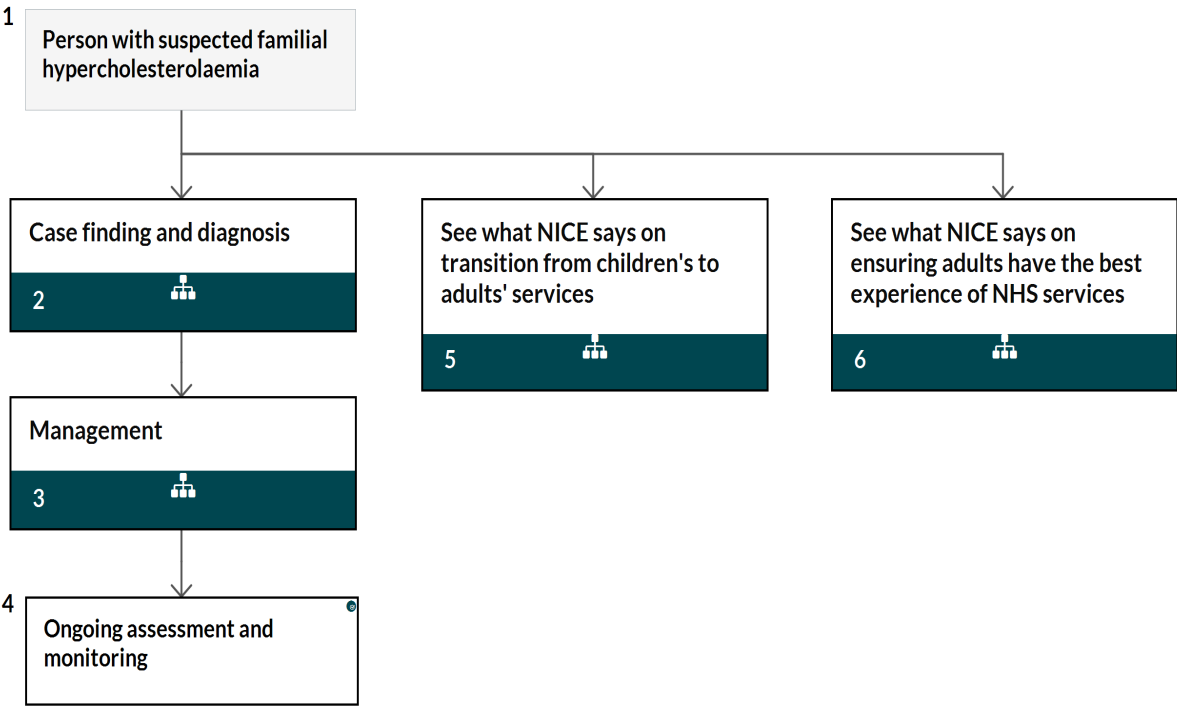
NICE Pathways bring together everything NICE says on a topic in an interactive flowchart. NICE Pathways are interactive and designed to be used online.

They are updated regularly as new NICE guidance is published. To view the latest version of this NICE Pathway see:

<http://pathways.nice.org.uk/pathways/familial-hypercholesterolaemia>

NICE Pathway last updated: 04 October 2019

This document contains a single flowchart and uses numbering to link the boxes to the associated recommendations.



## 1 Person with suspected familial hypercholesterolaemia

No additional information

## 2 Case finding and diagnosis

See Familial hypercholesterolaemia / Familial hypercholesterolaemia: case finding and diagnosis

## 3 Management

See Familial hypercholesterolaemia / Managing familial hypercholesterolaemia

## 4 Ongoing assessment and monitoring

All people with FH should be offered a regular structured review that is carried out at least annually.

A baseline ECG should be considered for adults with FH.

Healthcare professionals should record the progress of cascade testing among the relatives of a person with FH as part of the structured review. This should include at least the first-degree relatives and second-degree relatives and, when possible, third-degree relatives. If there are still relatives who have not been tested, further action should be discussed.

Healthcare professionals should update the family pedigree [See page 5] of a person with FH and note any changes in the coronary heart disease status of their relatives as part of the structured review. This should include at least the first- and second- and, when possible, third-degree biological relatives. [2008]

Structured review should include assessment of any symptoms of coronary heart disease and smoking status, a fasting lipid profile, and discussion about concordance with medication, possible side effects of treatment the patient may be experiencing, and any changes in lifestyle or lipid-modifying drug therapy that may be required to achieve the recommended LDL-C concentration.

## Quality standards

The following quality statement is relevant to this part of the interactive flowchart.

### 8. Annual review

#### **5 See what NICE says on transition from children's to adults' services**

[See Transition from children's to adults' services](#)

#### **6 See what NICE says on ensuring adults have the best experience of NHS services**

[See Patient experience in adult NHS services](#)

## Pedigree

A method of characterising the relatives of an index individual/case and their family relationship as well as problems or illnesses within the family. This information, often represented graphically as a family tree, facilitates analysis of inheritance patterns. Study of a trait or disease begins with the affected person (the index individual). The pedigree is drawn as the relatives are described. One begins with the siblings of the index individual and proceeds to the parents; relatives of the parents, including brothers, sisters, nephews, nieces, grandparents, and so on. At least three generations are usually included. Illnesses, hospitalisations, causes of death, miscarriages, abortions, congenital anomalies, and any other unusual features are recorded.

## Glossary

### ACE inhibitors

angiotensin-converting enzyme inhibitors

### Adults with FH

(for the purposes of this guidance, 'adults' includes all persons with familial hypercholesterolaemia (FH; heterozygous or homozygous) who are 16 years and older)

### Cascade testing

(a mechanism for identifying people at risk of a genetic condition by a process of family tracing; for FH the test employed is a DNA test where a disease-causing mutation has been identified in the index individual)

### Child-focused setting

(Child-focused refers to valuing the child's view and validating their voice in making decisions impacting their lives; a child-focused facility or space is one designed from the viewpoint of the service recipients)

### COCs

combined oral contraceptives

**Coronary heart disease**

(an event is defined as angina, acute coronary syndrome, myocardial infarction, need for coronary artery bypass grafting, need for percutaneous coronary intervention or definite coronary artery disease on coronary angiography)

**CVD**

cardiovascular disease

**ECG**

electrocardiogram

**Family history**

(the structure and relationships within the family that relates information about diseases in family members)

**FH**

familial hypercholesterolaemia

**First-degree relative**

(a person's biological parents, brothers and sisters, and children)

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**Heterozygous FH**

(high LDL-C concentration in the blood caused by an inherited mutation from one parent only)

**High-intensity statin**

(statins are classified as high intensity if they produce greater LDL-C reductions than simvastatin 40 mg (for example, simvastatin 80 mg and appropriate doses of atorvastatin and rosuvastatin))

**Index individual**

(the original patient who is the starting point for follow-up of other members of a family when investigating for possible causative genetic factors of the presenting condition; synonymous with proband)

**LDL**

low-density lipoprotein

**LDL-C**

low-density lipoprotein cholesterol

**Mutation**

(an identified change in the DNA sequence of a gene that is predicted to damage the normal function of the gene and so cause disease)

**Secondary causes of hypercholesterolaemia**

(causes of hyperlipidaemia other than familial, including uncontrolled diabetes mellitus, obesity, excess alcohol consumption, untreated hypothyroidism and some medications, for example, thiazide diuretics and ciclosporin)

**Second-degree relatives**

(a person's biological grandparent, grandchild, uncle, aunt, niece, nephew, half sister or half brother)

**Tendon xanthomata**

(a clinically detectable nodularity and/or thickening of the tendons caused by infiltration with lipid-laden histiocytes (macrophages in connective tissue); a distinctive feature of FH that most frequently affects the Achilles tendons but can also involve tendons on the back of the hands, elbows and knees)

**Third-degree relatives**

(a person's biological great grandparent, great grandchild, great aunt, great uncle, first cousin,

grand nephew or grand niece)

### **Urgent referral**

(as soon as possible with a maximum of 14 days)

### **Sources**

Familial hypercholesterolaemia: identification and management (2008 updated 2019) NICE guideline CG71

### **Your responsibility**

#### **Guidelines**

The recommendations in this guideline represent the view of NICE, arrived at after careful consideration of the evidence available. When exercising their judgement, professionals and practitioners are expected to take this guideline fully into account, alongside the individual needs, preferences and values of their patients or the people using their service. It is not mandatory to apply the recommendations, and the guideline does not override the responsibility to make decisions appropriate to the circumstances of the individual, in consultation with them and their families and carers or guardian.

Local commissioners and providers of healthcare have a responsibility to enable the guideline to be applied when individual professionals and people using services wish to use it. They should do so in the context of local and national priorities for funding and developing services, and in light of their duties to have due regard to the need to eliminate unlawful discrimination, to advance equality of opportunity and to reduce health inequalities. Nothing in this guideline should be interpreted in a way that would be inconsistent with complying with those duties.

Commissioners and providers have a responsibility to promote an environmentally sustainable health and care system and should assess and reduce the environmental impact of implementing NICE recommendations wherever possible.



## Technology appraisals

The recommendations in this interactive flowchart represent the view of NICE, arrived at after careful consideration of the evidence available. When exercising their judgement, health professionals are expected to take these recommendations fully into account, alongside the individual needs, preferences and values of their patients. The application of the recommendations in this interactive flowchart is at the discretion of health professionals and their individual patients and do not override the responsibility of healthcare professionals to make decisions appropriate to the circumstances of the individual patient, in consultation with the patient and/or their carer or guardian.

Commissioners and/or providers have a responsibility to provide the funding required to enable the recommendations to be applied when individual health professionals and their patients wish to use it, in accordance with the NHS Constitution. They should do so in light of their duties to have due regard to the need to eliminate unlawful discrimination, to advance equality of opportunity and to reduce health inequalities.

Commissioners and providers have a responsibility to promote an environmentally sustainable health and care system and should assess and reduce the environmental impact of implementing NICE recommendations wherever possible.

## Medical technologies guidance, diagnostics guidance and interventional procedures guidance

The recommendations in this interactive flowchart represent the view of NICE, arrived at after careful consideration of the evidence available. When exercising their judgement, healthcare professionals are expected to take these recommendations fully into account. However, the interactive flowchart does not override the individual responsibility of healthcare professionals to make decisions appropriate to the circumstances of the individual patient, in consultation with the patient and/or guardian or carer.

Commissioners and/or providers have a responsibility to implement the recommendations, in their local context, in light of their duties to have due regard to the need to eliminate unlawful discrimination, advance equality of opportunity, and foster good relations. Nothing in this interactive flowchart should be interpreted in a way that would be inconsistent with compliance with those duties.

Commissioners and providers have a responsibility to promote an environmentally sustainable health and care system and should assess and reduce the environmental impact of implementing NICE recommendations wherever possible.